

Opis choroby *

Definicja

Curly hair-acral keratoderma-caries syndrome is an extremely rare ectodermal dysplasia syndrome characterized by premature loss of curly, brittle, dry hair, premature loss of teeth due to caries, nail dystrophy with thickening of the finger- and toe-nails, acral keratoderma and hypohidrosis. Additionally, sparse eyebrows and eyelashes, receding frontal hairline and flattened malar region are associated. The severity of features appears to increase with age.

Dane

Klasyfikacja

Choroba

Synonimy

CHAC syndrome

CHACS

Zespół CHAC

CHACS

Kod ORPHA

307766

Kod OMIM

607656

Kod ICD10

Q82.8

Kod ICD11

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*Źródło

orphanet